



“Genetic counseling in CHARGE syndrome: Diagnostic evaluation through follow up”

By Meg Hefner and Emily Fassi [<https://doi.org/10.1002/ajmg.c.31589>]

AUTHORS AND THEIR CONNECTION TO THE CHARGE SYNDROME FOUNDATION:



Meg Hefner, MS, is a genetic counselor at Saint Louis University School of Medicine with 35 years experience with CHARGE syndrome (CS). She is a founding member of the CHARGE Syndrome Foundation and continues to serve as an Advisor to the Board. Meg has written a myriad of publications for professionals and families, including the Management Manual for Parents and (as co-editor) the 2011 book on CHARGE syndrome [Plural Publishing]. In addition to creating a clinical database to share CHARGE information with other researchers, Meg coordinates the research done at conference and is a mentor to the Sandra Davenport CHARGE Syndrome Fellows.



Emily Fassi, MS, is a genetic counselor who has been involved with CHARGE since before her training as a genetic counselor. She helped develop the CHARGE Syndrome Clinical Database Project (CSCDP) with Meg Hefner beginning in 2011. Emily received a scholarship to attend the 2013 Conference and attended the 2017 Conference as a Davenport Fellow (young professional) where she presented a poster on developmental milestones in CHARGE. She currently works as a cancer genetic counselor in Boise, Idaho and continues to have a major interest in CHARGE syndrome and deafblindness.

SUMMARY OF THE PAPER:

CHARGE syndrome (CS) is a complex genetic disorder causing multiple birth defects and sensory deficits (hearing, vision, balance, smell). Genetic counseling in CS must include not only the provision of factual information about CS, its cause, and inheritance, but also information about the developmental implications of CS features, referral to appropriate resources, and assistance with understanding this information. CS should be considered in patients with any of the major diagnostic features: coloboma, choanal atresia, semicircular canal anomalies, or cranial nerve anomalies. The most common other conditions with features that overlap with CHARGE are 22q11.2 deletion and Kabuki syndromes. Evaluation of features and what the children look like, along with genetic testing, can usually distinguish between the three syndromes. Genetic counseling is important from early on, to help the family understand the process of genetic diagnosis, to interpret information coming from other specialists and to provide support and resources. Parents can easily be overwhelmed with the complexity of issues facing their child at diagnosis and in the future. CS is a substantial burden on a child, with high early mortality, multiple illnesses, hospitalizations and surgeries, and apparent medical fragility throughout life. The medical complexity of CS disrupts family life and contributes to the delays in development.

Multiple sensory deficits (impaired vision, hearing, and balance) further contribute to delayed motor and language development despite many individuals with CS having normal intelligence. Early referral to specialists in deafblindness and sensory deficits is essential. Resources are available to assist genetic counselors in diagnosis, follow-up, and management of patients with CS.

Tables & Figures in the paper include: Clinical diagnostic criteria; Photos of faces, hands, ears; What else looks like CHARGE; Resources for genetics team and families; Recurrence risks.

WHAT DOES THIS MEAN TO FAMILY/PERSON WITH CHARGE?

This paper covers the ideal of what genetics evaluation and follow up would entail for a child with possible CHARGE. If you have questions about how a diagnosis is made, what else looks like CHARGE, or the chance of it happening again, the tables may be helpful. The list of resources may be helpful to you and/or your genetics team.

SHOULD I READ IT? SHOULD ONE OF MY DOCTORS READ IT?

Yes to both. It is written at a level that should be understandable to both parents and professionals. It brings together information that might otherwise take a lot of time to pull together. This paper is a good summary to help explain parts of CHARGE to your primary physician, friends and family.

FULL CITATION:

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